Instructions: *Use Times New Roman font size 11, 1.0 pt text space, and* ***maximum three A4 pages****, including any figures.*

Name or type of instrument	Bionano genomics: Saphyr Optical gene mapping
Unit	Clinical Genomics Linköping
Platform	Clinical Genomics
Applicant (Head of Unit)	Tobias Strid
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Instrument, acquisition cost (kSEK)	2400
Funding applied for (kSEK)	2400

Description of the instrument

Describe the instrument, its purpose, and its specific capabilities. If possible, provide a link to a web page describing the type of instrument. Does the instrument represent a new technology at the unit, or is it a replacement/upgrade of current equipment base? Is the instrument unique in terms of service to users nationally?

The Bionano genomics: Optical gene mapping (OGM) Bionano genomics instrument (https://bionanogenomics.com/, https://bionanogenomics.com/clinical/cytogenomics/) bridges the information gap between whole genome sequencing, microarray analysis and traditional karyotyping. It detects broken, missing, fused and rearranged genes, or extra chromosomes and is able to diagnose disease and define treatment options. Compared to traditional cytogenetic methods, the Bionano OGM has a much greater resolution and can define breaking points more accurately than earlier methods. OGM is able to detect all CNVs, chromosomal aberrations and structural variants. Except being an alternative to traditional molecular methods, it is also able to detect genetically complex aberrations where complementary, often more complex and expensive, methods otherwise would be necessary, e.g. Long Read sequencing. Scanning and data-generation is performed in the OGM instrument and ready for analysis and interpretation on the same day. 3 paralleled human genomes are scanned in 6h.

The method and its associated laboratory process is rapid and easy. Starting with DNA isolation at mega-base size followed by enzymatic labeling and transfer of labeled DNA into cartridges for scanning.

The instrument will be an addition of a new technology to the Linköping node and a great addition to the total capacity to CG to perform OGM and to provide national coverage, since the instrument at present is available only at one node.

Statement of need

Describe how the instrument will be crucial for the operations, productivity, development, and competitiveness of your unit. Estimate the need and impact of the instrument from a user perspective. Will the instrument be shared with or used by other units or research groups at SciLifeLab? Also, describe any co-funding plans.

Genetic karyotyping is a central part of medical genetics, both for research questions and diagnostics. Complex rearrangements and gene fusions are also gaining more importance in cancer biology. The methods used today are generally highly manual, labor intensive and less precise then OGM. Competence and equipment handling the type of questions dealt with using OGM is currently centered at the Stockholm/Uppsala region, hence an establishment of this technique in Linköping would greatly expand the overall availability and could establish a hub for the southern part of Sweden and a national center for analysis and care of neuromuscular disorders and highly specialized care of e.g. Facioscapulohumeral muscular dystrophy, thus making a direct link to the clinic and health care. Optical Genome Mapping constitutes one of the current focus areas of the Clinical Genomics platform. The focus area is currently run from CG Stockholm, but the addition of this instrument will enable CGLi to join forces and complement in this cross-node task, as well as stimulate genomic research on other CG focus areas.

The University hospital in Linköping has been assign by The National board of Social Affairs and Health ("Socialstyrelsen") as a center for national highly specialized care related to neuro muscular disorders. These disorders are often caused by structural variants that can be detected by OGM. The instrument in question is also able to detect diseases caused by genomic expansions. One example of such a disease is Facioscapulohumeral muscular dystrophy that presently is analyzed using time consuming and hazardous Southern blot. The care and diagnostics part of the program for national highly specialized care is only allowed to be offered at designated sites, associated with clinical efforts but will most likely also attract clinical research within the field, since a close connection to clinical practice is often a prerequisite. Hence, an OGM will be a key instrument addition to the infrastructure in Clinical Genomics Linköping to aid in transfer of state of art technology to health care which is one of the main missions of the Clinical Genomics Platform.

Users: Instrumentation for optical genome mapping has a high potential of national customers since the current availability of such instrumentation is rare. There are several potential local/regional research and clinical customers both at Linköping University and at Region of Östergötland which will greatly aid in initial development of the technology.

Co-founding: We will apply for co-founding also within the ALF system, which constitutes the basis for instrument investments in our local core facilities in Linköping.